ABSTRACT

Significance of MLL gene aberrations in patients with acute myeloid leukemia

In acute myeloid leukemia (AML), predominantly in AML M5a, the most frequent recurrent aberration of chromosome 11 involves region 11q23. Molecular breakpoint studies of several translocations involving chromosomal band 11q23 led to the detection of a gene that was named MLL (**m**yeloid/**l**ymphoid **l**eukemia). Since that time, more than 70 different translocation partners of the MLL gene have been described. This gene is important for the proper HOX gene expression during ontogenesis and hematopoiesis. Chromosomal aberrations affecting the MLL gene occur in 5 - 10 % of AML cases and are very variable. Aberrations of the MLL gene are associated with an aggresive type of the disease and its detection is needed for the treatment decision. Therefore, we investigated the occurrence of MLL abnormalities in bone marrow cells of the 66 newly diagnosed AML patients, using conventional cytogenetic and fluorescence in situ hybridization (FISH) analyses with a commercially available MLL Break Apart Rearrangement probe (Abbott VYSIS). Out of the 66 patients, we proved MLL abnormalities in 9 (13,6%): 5 (7,6%) showed translocation of MLL gene, in 3 (4,5%) we detected MLL gene amplification without any evidence of rearrangement and in 1 (1,5%) pacient only an extra copy of the MLL gene. The FISH results were verified by multicolor FISH (mFISH) and multicolor banding (mBAND). In this study, we firstly described new MLL gene fusion partner - gene TEL (**t**ranslocation **e**ts **l**eukemia, 12p13).

Key words: acute myeloid leukemia, MLL, translocation, amplification, complex rearrangement of the karyotype, FISH, TEL.

Klíčová slova: akutní myeloidní leukemie, MLL, translokace, amplifikace, komplexní přestavby karyotypu, FISH, TEL.